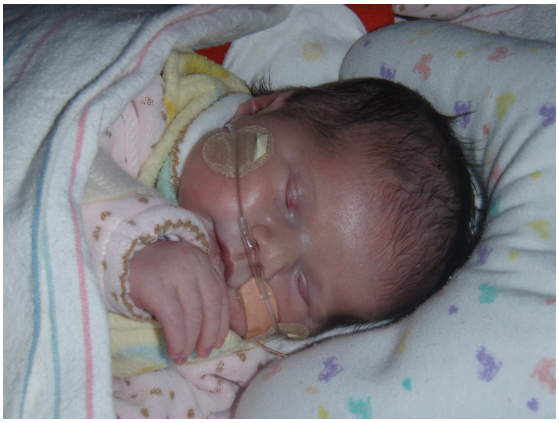

TRIS project
2014

Mackenzie was born in 2009 and was diagnosed three days after birth (trisomy 9 mosaic). She lives in the United States. Participation in the TRIS project began in 2013 at the age of 44 months.

Mother was 39 and father was 37 years old at time of conception. After conception, parents reported general knowledge of rare trisomy conditions. Child with rare trisomy was the second of two pregnancies (gravida, 2; para, 2). Older sibling has been diagnosed with an autism spectrum disorder.

Birth Information

Female infant was delivered via emergency caesarian section at 39+5 weeks gestation. Infant was born weighing 3020 grams and 46 centimeters in length in November 2009. APGAR scores at one and five minutes were 2 and 2, respectively and 8 at ten minutes. Infant was identified with larger than typical sized ventricles in the brain prior to delivery.

At birth, infant presented with low ears, small jaw, small & sunken appearing eyes, short palpebral fissures small anteverted nostrils, widely arched palate, thumbs with tightness to abduction, small chest, deep sacral dimple, dorsiflextion feet with limited plantarflexion, torticollis, increased generalized tone. Infant also presented with the following medical issues: respiratory difficulties, heart murmur, patent ductus arteriosis (PDA), ventricular septal defect (VSD), aberrant left subclavian artery, hypocalcemia, thrombocytopenia, congenital hypothyroidism (continues to be treated with Levothyroxine), feeding difficulties, and jaundice. Infant was diagnosed with trisomy 9 mosaic three days after birth.

Infant received the majority of her post-birth care in a specialized hospital's neonatal intensive care unit (NICU). Infant was on the unit for 3.5 weeks. Infant was also on a respirator and a ventilator for first 24 hours after birth. Oxygen was delivered via nasal cannula after first 24 hours. A heart monitor and pulse oxymeter were also used prior to discharge. Infant was fed via IV feedings, nasogastric tube (n-g tube) and, later, after surgery, gastrostomy tube (g-tube).

An audiology exam, echocardiogram, ophthalmology exam, ultrasound, x-ray series and blood transfusion were also performed in the post-birth care unit. Infant's feeding difficulties where repaired through placement of a gastrostomy feeding tube 18 days after birth. Discharge weight was 3165 grams.

Infant received palliative care after discharge from the post-birth unit. Palliative care was discontinued at age three after feeding was stabilized.

Surgeries

Infant's feeding difficulties (oral motor dysfunction and aspiration) were addressed 18 days after birth with placement of a percutaneous endoscopic gastrostomy tube (PEG g-tube). This type of g-tube was surgically replaced with a Mic-Key Button at the age of six months.

Also, at six months of age, child had received ear tubes for otitis media (recurrent middle ear infections). At seven months after birth, infant had surgery to correct dyspasia of right hip and adductory longus for left hip contracture. A spica cast was needed for eight weeks and a brace for the following six weeks. To address obstructive apnea, infant was treated with two surgical procedures: adenoidectomy and tonsillectomy at 26 months.

Child's strabismus was corrected at 29 months.

No surgeries were required for cardiac conditions.

Cardiac

Infant presented with a heart murmur, PDA, VSD, and aberrant left subclavian artery. No surgical interventions were necessary to correct these conditions. All heart conditions resolved.

Respiratory

Prior to discharge from the NICU, infant required oxygen, respirator, and ventilator assistance but did not need these items after discharge. Child is also noted to have micrognathia.

At nine months, Pulmicort and Albuterol were prescribed to prevent pneumonia. Child still uses these prescriptions as needed application with a nebulizer. Airborne allergies were reported in the follow-up survey at 56 months and treated with diphenhydramine HCI oral solution.

At 26 months of age, child required removal of adenoids and tonsils to prevent obstructive apnea. Mother reports loud snoring was the only symptom of this condition. Sleep apnea was never diagnosed.

Neurological

Mother reports seizures due to obstructive apnea shortly after child was born lasting for approximately 24 hours. No medications were necessary to treat this condition. Child was moved from supine to prone. Seizures stopped with the change in positioning allowing chin to move forward to counteract infant's jaw moving backward and cutting off her airway.

Renal

No renal conditions reported in baseline data. Follow-up data indicates constipation as a concern but no treatment is indicated. More free water needed in diet.

Gastrointestinal

Child was diagnosed with gastrosophageal reflux (GER) and aspiration while still in the NICU. Condition continues to be effectively treated with Zantac.

Nutrition

Immediately after birth, infant was fed primarily by nasogastric tube (n-g tube). Infant was not permitted to latch on to be fed by bottle or breast due to aspiration issues. A gastronomy tube was surgically placed 18 days after birth.

At time of baseline survey, child received g-tube feedings primarily. At Year 1 Follow-up, child continued to receive g-tube feedings as well as pureed foods orally. Child has also passed a swallow study allowing for additional oral feeding to start. A gastroenterologist is involved in the child's care.

Oral health

Child began dental visits at the age of 27 months with a dentist specializing in working with children. Child requires sedation for routine dental care. Mother reports child has excessive plaque, due to lack of oral feeding and GER. Mother notes that plaque is diminishing as more oral stimulation is provided and intake increases.

Vision

Child was diagnosed with strabismus at approximately nine months. This condition was surgically corrected at 29 months.

Child's initial vision-related diagnosis was intermittent alternating exotropia at 32 months. At 38 months of age, diagnoses of optic atrophy ("dull" optic nerve), hypermetropia and astigmatism were made when child were added. Hypertropia was identified at 44 months. An ophthalmologist follows the remaining conditions at six-month visits. Glasses are not needed at this time.

Auditory

Child has a documented hearing loss (mild to moderate in left ear; wears a hearing aid) based on auditory brainstem response (ABR) results. In addition, otitis media was first diagnosed at one month of age. Child had tubes placed at five and 26 months. The latter was during tonsil and adenoid surgeries. Child has also used hearing aids since 18 months of age as well as American Sign Language.

Immunizations

Immunizations began and continued at the ages recommended by the American Association of Pediatrics for Diptheria, Tetanus and Pertussis (DPT), Haemophilus influenzae type B (HIB), Hepatitis A (HepA), Hepatitis B series, Inactive polio virus, Influenza, Measles, Mumps and Rubella (MMR), Pneumococcal pneumonia, Respiratory syncytial virus (RSV) and Varicella. No adverse reactions are noted.

Orthopedic

At birth child presented with torticollis, thumb in palm, hip dislocation, and tightness of dorsal structures bilaterally at ankles and feet. At seven months, infant had surgery to correct dyspasia of right hip and adductory longus for left hip contracture.

Child was also diagnosed with scoliosis and kyphosis at 3.5 months. Both conditions are being monitored at this time with no immediate plans for surgical intervention. An orthopedic surgeon monitored the child at six month intervals for the first three years and, now, yearly.

Child has used McKie hand splints for approximately the first seven months after birth and ankle foot orthotics until age four. After child's fourth birthday, a change was made to a foot orthotic with flexible metal shoe insert to providing bracing to calf.

Developmental milestones

At completion of baseline survey at 44 months of age, child demonstrated most 12-month milestones including standing, demonstrating preference for toys/objects, and using two hands to explore objects. In addition, child was able to scribble with a crayon, sort objects based on one characteristic (e.g., size), use signs and gestures to communicate needs, attempt to throw or kick a ball and play cooperatively with sibling (a 36 month skills).

At follow-up at 56 months, child was able to walk independently. Child is using American Sign Language to communicate wants and needs to her parents and teachers. Child was also better tolerating oral stimulation and accepting more food orally.

Developmentally, child uses sign language to communicate (approximately 36 signs) and vocalizes sounds during music activities at preschool (e.g., mama, baba). When presented with two songs or preferred items, child demonstrates choice making. Child enjoys playing with toys with lights and music as well as sensory play activities including painting. Fine motor skills such as approximations of age appropriate grasps and copying vertical and horizontal lines using a slant board are improving. Child is able to interact with apps on an iPad. Finally, child is showing increased interest in same age peers including observing them during small group activities.

Child participates in indoor and outdoor gross motor activities with adult support including playground equipment and an adapted tricycle. Mother notes child especially enjoys climbing on various play structures. Child is also noted to be cooperative during dressing activities. Child can also complete some steps of hand washing with little to no adult assistance.

Education and therapy services

Child received early intervention services up to 36 months of age including nursing and nutrition services. In addition, audiology and vision services were provided. Occupational, physical and therapy services were offered as well. Developmental therapy was also provided. A developmental pediatrician was also involved in child's care.

After the age of three, child received services in both a general education and special education classroom in a public school.

At baseline survey completion, child was receiving 45 minutes of occupational therapy weekly at a clinic. Physical and speech therapy were each provided for 90 minutes per week also in a clinic setting. Child has used a gait trainer as well.

At present, child is in an integrated preschool classroom four half days a week (2.5 hours per day) and continues to receive physical, occupational and speech therapy services with twice weekly 45-minute session privately. Therapy services are also provided at child's public school

program (once a week for each type of therapy). Child receives deaf and hard of hearing therapy services in the preschool classroom. Child is also involved with a local special needs T-ball league.

For more information on the Tracking Rare Incidence Syndromes (TRIS) project:

Homepage: http://web.coehs.siu.edu/grants/tris/

Case studies page: http://web.coehs.siu.edu/Grants/TRIS/casestudies.html

Facebook page: https://www.facebook.com/TRIS.Trisomy.project